



RNASEH2B gene

ribonuclease H2 subunit B

Normal Function

The *RNASEH2B* gene provides instructions for making one part (subunit) of a group of proteins called the RNase H2 complex. This complex is a ribonuclease, which means it is an enzyme that helps break down RNA, a chemical cousin of DNA. In particular, the RNase H2 complex helps break down molecules in which one strand of RNA is combined with one strand of DNA (RNA-DNA hybrids). These hybrids are formed during DNA copying (replication) and are found in all cells.

The RNase H2 complex is involved in DNA replication, error repair, and other cellular functions. Researchers believe that these additional functions may include helping to prevent inappropriate immune system activation.

Health Conditions Related to Genetic Changes

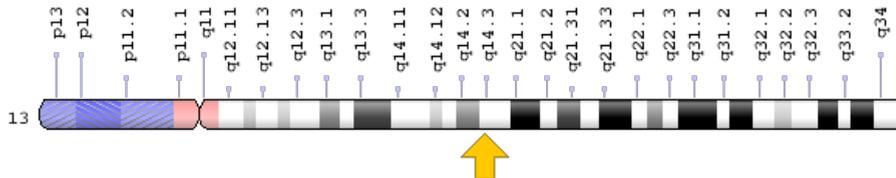
Aicardi-Goutieres syndrome

At least 20 mutations in the *RNASEH2B* gene have been identified in people with Aicardi-Goutieres syndrome. These mutations likely result in a dysfunctional RNase H2 complex. The resulting disruption in cellular functions may lead to an accumulation of unneeded DNA and RNA in cells. These DNA and RNA molecules or fragments may be generated during the first stage of protein production (transcription), replication of cells' genetic material in preparation for cell division, DNA repair, cell death, and other processes. The unneeded DNA and RNA may be mistaken by cells for those of viral invaders, triggering immune system reactions that cause severe brain dysfunction (encephalopathy), skin lesions, and other signs and symptoms of Aicardi-Goutieres syndrome.

Chromosomal Location

Cytogenetic Location: 13q14.3, which is the long (q) arm of chromosome 13 at position 14.3

Molecular Location: base pairs 50,909,678 to 50,970,462 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGS2
- Aicardi-Goutieres syndrome 2 protein
- deleted in leukemia 8 protein
- deleted in lymphocytic leukemia 8
- DLEU8
- FLJ11712
- ribonuclease H2 subunit B isoform 1
- ribonuclease H2 subunit B isoform 2
- ribonuclease H2, subunit B
- ribonuclease H1 subunit B
- RNase H2 subunit B
- RNH2B_HUMAN

Additional Information & Resources

GeneReviews

- Aicardi-Goutieres Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1475>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28RNASEH2B%5BTIAB%5D%29+OR+%28%28AGS2%5BTIAB%5D%29+OR+%28FLJ11712%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- RIBONUCLEASE H2, SUBUNIT B
<http://omim.org/entry/610326>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RNASEH2B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RNASEH2B%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25671
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/79621>
- UniProt
<http://www.uniprot.org/uniprot/Q5TBB1>

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